Published in final edited form as:

Am J Med Genet A. 2013 January ; 161A(1): 70–75. doi:10.1002/ajmg.a.35682.

Reproductive Patterns Among Mothers of Males Diagnosed With Duchenne or Becker Muscular Dystrophy

Sarah K. Nabukera[1], **Paul A. Romitti**[1,*], **Kristin M. Caspers**[1], **Natalie Street**[2], **Christopher Cunniff**[3], **Katherine D. Mathews**[4], **Deborah J. Fox**[5], **Soman Puzhankara**[1], **Emma Ciafaloni**[6], **Katherine A. James**[7], **Yin Su**[5], and **the MD STAR*net***

[1]Department of Epidemiology, College of Public Health, The University of Iowa, Iowa City, Iowa

[2]National Center on Birth Defects and Developmental Disabilities, Centers for Disease Control and Prevention, Atlanta, Georgia

[3]Department of Pediatrics, College of Medicine, The University of Arizona, Tucson, Arizona

[4]Departments of Pediatrics and Neurology, Carver College of Medicine, The University of Iowa, Iowa City, Iowa

[5]The Congenital Malformations Registry, Bureau of Environmental and Occupational Epidemiology, Center for Environmental Health, New York State Department of Health, Troy, New York

[6]Department of Neurology, School of Medicine and Dentistry, University of Rochester, Rochester, New York

[7]Division of Disease Control and Environmental Epidemiology, Colorado Department of Public Health and Environment, Denver, Colorado

Abstract

Diagnosis of a child with Duchenne or Becker muscular dystrophy (DBMD) may impact future maternal reproductive choice; however, little is known about the reproductive patterns of mothers with a male child diagnosed with DBMD. Using population-based surveillance data collected by the muscular dystrophy surveillance, tracking, and research network, the proportion of mothers who conceived and delivered a live birth following the diagnosis of DBMD in an affected male child and factors associated with such reproductive choice were identified. To accomplish this, maternal demographic data were linked to birth certificate data to construct the reproductive history for 239 mothers. Univariable and bivariable analyses were conducted to determine the proportion of mothers delivering a live birth and associated factors. By the time of the current study, 96 (40.2%) of the 239 mothers had at least one live birth following delivery of their oldest

[*]Correspondence to: Paul A. Romitti, Ph.D., Associate Professor, Department of Epidemiology, The University of Iowa, College of Public Health, S416 CPHB, 105 River St., Iowa City, IA 52242-5000. paul-romitti@uiowa.edu.

The authors have no conflict of interest to declare.

Disclaimer: The findings and conclusions in this report are those of the authors and do not necessarily represent the official position of the Centers for Disease Control and Prevention.

Findings reported in this paper were presented at the National Birth Defects Prevention Network (NDBPN) 14th Annual Meeting, Orlando, FL; February 28–March 2, 2011.

affected male child; 53 (22.2%) of these mothers had a live birth before and 43 (18.0%) had a live birth after DBMD diagnosis of a male child. Mothers with a live birth after diagnosis were significantly younger at diagnosis of the oldest affected male child (26.2 ± 4.2 years vs. 31.5 ± 5.5 years), and were less likely to be white non-Hispanic compared to those with no live birth after diagnosis. These results suggest that about one in five mothers deliver a live birth subsequent to DBMD diagnosis in a male child. Maternal age and race/ethnicity were associated with this reproductive choice.

Keywords

Becker muscular dystrophy; Duchenne muscular dystrophy; live birth; pregnancy

INTRODUCTION

Duchenne (OMIM 310200) and Becker (OMIM 300376) muscular dystrophies (DBMD) are X-linked progressive muscle conditions, affecting approximately 1.3–1.8/10,000 males [Centers for Disease Control and Prevention (CDC), 2009]. The primary mode of genetic transmission for these conditions is due to the mother being a carrier of an altered dystrophin gene, and the remainder is due to a new mutation in the affected son; thus, mothers of males with DBMD may or may not have a family history of these conditions. Early signs and symptoms of DBMD include difficulty walking, running, or climbing stairs, and progressive muscle weakness. Currently, no cure exists for DBMD. Management includes use of corticosteroids to slow disease progression and treatment of associated cardiac, pulmonary, and skeletal complications [Bushby et al., 2010a,b].

Diagnosis of DBMD in a child may influence a mother's reproductive choices; however, only two published investigations have examined the reproductive patterns of mothers following the diagnosis of Duchenne muscular dystrophy (DMD) in a male child. One investigation, a pilot study of eight mothers of males diagnosed with DMD by newborn screening and three female relatives at high risk of being carriers, found that four (50%) of the eight mothers with an affected male child had one or more subsequent pregnancies [Hildes et al., 1993]. The second investigation, a pilot study of 20 families with an affected male child with DMD identified by newborn screening, found that four (20%) families chose not to have another pregnancy after the diagnosis and 11 (55%) opted to delay additional pregnancies [Parsons et al., 2002]. Additional data suggest that males born to women with a prior knowledge of DMD family history are often diagnosed at an earlier age [Gardner-Medwin et al., 1978; Smith et al., 1989] compared to males born to women with no such knowledge [Ciafaloni et al., 2009]. Given this preliminary evidence, data from the population-based muscular dystrophy surveillance, tracking, and research network (MD STAR*net*) were used to examine reproductive patterns among mothers of males diagnosed with DBMD. Specifically, the proportion of mothers who conceived and delivered a live birth following the diagnosis of DBMD in an affected male child was calculated, and factors associated with such reproductive choice were examined.

MATERIALS AND METHODS

Study Design and Setting

A retrospective cohort study using data collected by the MD STAR*net* was conducted. The MD STAR*net* is a multisite project established in 2002 by the Centers for Disease Control and Prevention (CDC) to conduct population-based surveillance for individuals with DBMD born on or after January 1, 1982, and who resided in an MD STAR*net* site. Initially, the MD STAR*net* was comprised of four sites, Arizona (AZ), Colorado (CO), Iowa (IA), and western New York State (NY). Subsequently, activities were expanded to include Georgia (GA) in 2006 and Hawaii (HI) in 2008. Details of this project have been described elsewhere [Miller et al., 2006].

Study Population and Sample

The population for this study was comprised of mothers of males with a characteristic clinical course for DBMD defined by an elevated creatine kinase (CK) level, and either a dystrophin analysis that showed apathogenic abnormality, amuscle biopsy that showed abnormal dystrophin by immunostaining or Western blot, or a documented family history of an X-linked muscular dystrophy [Mathews et al., 2010]. Surveillance data collected through 2008 were used for this study, and included data on 536 mothers from AZ, CO, GA, IA, and NY with affected males born from 1982 to 2006; surveillance had not yet commenced in HI. Of the 536 mothers, 218 from CO and GA were excluded because birth certificate (BC) data were not available for linkage, and seven mothers from AZ, IA, and NY were excluded as the oldest affected male was adopted and BC data on the biological mothers were not available leaving 311 eligible mothers for data linkage and analyses.

Data Sources and Variables

Surveillance data were collected from medical records (MRs) and the BC, where available, for an affected male child, and included sociodemographics, birth history, clinical signs and symptoms, diagnostic tests, mobility, rehabilitation, co-morbidities (pulmonary, cardiac, skeletal, and psychosocial), and family history of DBMD. To construct maternal reproductive history subsequent to the birth of the oldest affected male child, MD STAR*net* data were linked to state BCs, through 2009. From the BC linkage, data were collected on birth date and sex of each subsequent live birth and maternal parity. A combination of deterministic and probabilistic approaches was used to match maternal MD STAR*net* and BC data [Romitti et al., 2010]. Institutional review boards from AZ, IA, and NY approved the study.

Independent variables examined to describe the study sample were: age (years) of the oldest affected male child at first creatine kinase (CK) testing (used as a proxy for DBMD diagnosis); maternal age (years), race/ethnicity (white non-Hispanic, Hispanic, black non-Hispanic, Native American, Asian or Pacific Islander, and other), and education (less than high school, high school, some college and college degree or higher); parity (0, 1 and 2 or more pregnancies); knowledge of DBMD family history (known history, no known history, and undetermined history) at delivery of her oldest affected male child; carrier status (carrier, suspected carrier, non-carrier, and unknown); and receipt of genetic counseling

(recommended, received, and unknown) at any time point prior to completion of surveillance data collection in 2008. To define knowledge of family history, MR and pedigree data were used to determine if the mother had a “known history” of other affected maternal relatives (e.g., an uncle, cousin, grandfather, or great uncle) diagnosed with DBMD, “no known history” of other affected family members, or “undetermined history” if data were insufficient to determine prior knowledge of DBMD family history. Carrier status was defined from pedigree data, maternal CK levels, and when available, DNA test results. A mother was classified as a “carrier” if the pedigree indicated she was an obligate carrier or if genetic testing detected a DBMD mutation; as a “suspected carrier” if the pedigree showed possible carrier status, and/or the mother had an elevated CK but genetic testing was unavailable; as a “non-carrier” if genetic testing indicated no dystrophin mutation in the mother; or as an “unknown carrier” if insufficient data were available to determine carrier status. Receipt of genetic counseling was defined from MR data, and was classified as “recommended” if counseling was recommended but there was no indication to suggest that it was completed; as “received” if counseling was completed; or as “unknown” if there were no data to indicate if counseling was recommended or received. Independent variables compared between mothers who did and did not deliver a live birth after diagnosis of their oldest affected male child included each variable listed above, except that maternal age was at the time of DBMD diagnosis of the male child, and parity was categorized (1, 2 and 3 or more pregnancies) to include, at a minimum, the delivery of the oldest affected male child.

A mother who conceived and delivered one or more live birth(s) after diagnosis of her oldest affected male child was categorized as having a live birth after diagnosis, and a mother who did not have a live birth or who had a live birth before diagnosis of her oldest affected male child was categorized as having no live birth after diagnosis. To determine if a mother had a live birth after diagnosis of her oldest affected male child, age at first CK for the child was compared to the birth intervals of each delivery that occurred after the birth of the child. To allow fora 3-month preconception interval and a term pregnancy, 1 year was subtracted from the birth interval of each subsequent live birth. If the birth interval was larger than the interval from birth to age at first CK, then a mother was classified as having a live birth after diagnosis of her oldest affected male child.

Data Analysis

Univariable and bivariable analyses were conducted to estimate frequencies and percentages for categorical variables and to estimate means, standard deviations (SDs), and ranges for continuous variables. The chi-square or Fisher exact tests, as appropriate, were calculated to assess significant differences ($P < 0.05$) for categorical variables, and *t*-tests were calculated to assess significant differences for continuous variables. Because of small numbers, education categories (less than high school, high school, some college, and bachelor’s degree or higher) were collapsed into high school or less, some college, and college degree or higher. Similarly, race/ethnicity categories (white non-Hispanic, Hispanic, black non-Hispanic, Native American, Asian or Pacific Islander, and other) were collapsed into white non-Hispanic and all other race/ethnicity. All analyses were conducted using Statistical Analysis Software (SAS) version 9.2, Cary, North Carolina, U.S.

RESULTS

Of the 311 mothers from AZ, IA, and NY identified for record linkage, 72 mothers were excluded from analyses producing a final analytic sample of 239 mothers. Mothers were excluded if: the oldest affected male child was born before 1982 (n = 4), was not identified in the 2008 surveillance data (n = 1), or had an unknown age at DBMD diagnosis (n = 12); BC data were missing (n = 43); or knowledge of DBMD family history at delivery of their oldest affected male child was undetermined (n = 12). Mothers included in the final analyses and those excluded differed by race/ethnicity, carrier status, and receipt of genetic counseling (Table I).

At the time of the current study, 96 (40.2%) of the 239 mothers had delivered one or more live birth(s) following the birth of their oldest affected male child. Fifty-three (22.2%) mothers delivered 63 live births (28 females and 35 males) before diagnosis of their oldest affected male child; 12 of the 35 males were identified by the MD STAR*net* as having DBMD. The remaining 43 (18.0%) mothers delivered 53 live births (20 females and 33 males) after diagnosis of their oldest affected male child with 12 of the 33 males identified by the MD STAR*net*. To evaluate potential differences in the opportunity to conceive between mothers who had no live birth and mothers who either had a live birth before or after diagnosis of their oldest affected male child, the intervals between maternal age at BC request and maternal ages at delivery and at diagnosis of their oldest affected male child were compared. No statistically significant differences were found (data not shown).

Table II shows maternal factors associated with having a live birth following the diagnosis of DBMD in an affected male child. Mothers who had a live birth after diagnosis of their oldest affected male child were more likely to be younger and less likely to be white non-Hispanic compared to mothers who did not have a live birth after diagnosis. Mothers in each group tended not to differ by: education and knowledge of DBMD family history at time of delivery of the oldest affected male child; parity at diagnosis of the oldest affected male child; or carrier status and receipt of genetic counseling at any time point prior to surveillance data collection in 2008.

DISCUSSION

Approximately 18% of mothers in this study had a live birth following the diagnosis of their oldest affected male child, which is lower than the 50% reported in one previously published pilot study [Hildes et al., 1993], but similar to the 25% reported in another pilot study whose reproductive choices were not affected by such diagnosis [Parsons et al., 2002]. Differences in study design, sample size, and study populations may explain the variability in findings. The two previous studies [Hildes et al., 1993; Parsons et al., 2002] were pilot projects that used samples sizes of 8 and 20 mothers, respectively, identified through newborn screening for DBMD, whereas the current study was based on a large sample of mothers of male children with DBMD identified through a population-based surveillance approach. Mothers of affected male children identified through newborn screening may be more likely to have a second child before their affected male child begins to show signs of the disease. Observing the progression of DBMD in their own child may have a greater effect on the mother's

future reproductive choices compared to knowing that there may be a chance that a future child may inherit the disease. The fact that knowledge of a positive family history did not differ in the current study between mothers who did and did not have another child following a diagnosis in an older child is consistent with this interpretation. As such, these mothers may have opted not to have another child. Other reasons that may explain the observed differences between this study and previous studies may be related to a general population decline in birth rates over time [Hamilton and Ventura, 2006] or use of reproductive technologies such as prenatal testing [Raymond et al., 2010], which may lead to termination of pregnancies with an affected fetus. The retrospective design of the current study did not permit the latter factor to be fully assessed.

Factors associated with reproductive decisions for mothers at risk of having a child with a genetic disorder have been examined previously. Published findings suggest that a mother's desired number of children [Hildes et al., 1993], receipt of genetic counseling [Abramovsky et al., 1980; Rona et al., 1994; Eggers et al., 1999; Meldrum et al., 2007], perception of disease risk [Hutton and Thompson, 1976; Abramovsky et al., 1980; Wertz et al., 1984], concerns about the effects of an affected child on one's social and personal life and problems caring for the child [Wertz et al., 1984; Frets et al., 1991], and disapproval of relatives [Frets et al., 1991], may play a role in decisions about reproduction.

In the current study, mothers' age at diagnosis of her oldest affected male child and her race/ethnicity were each associated with having a live birth following the diagnosis. The observed association of younger mothers having an increased tendency to have additional children could be a reflection of the general population trend of younger mothers having more births compared to older mothers [Hamilton et al., 2006]. Fertility rates are also higher in younger mothers [American Society of Reproductive Medicine, 2008]. Conversely, younger mothers may feel as though they have sufficient resources, both internally and externally, to have another child regardless of diagnosis. Cultural and/or religious differences with respect to reproduction may explain the findings related to race/ethnicity [Brotto et al., 2008; Maternowska et al., 2010]. Family pressures to have more children and cultural norms regarding family planning have been reported to influence reproductive choice [Godley, 2001; Oladeji, 2008]. Unfortunately, these differences could not be fully examined using the available MD STAR*net* data.

Parity at diagnosis of the oldest affected male was not associated with having a live birth after DBMD diagnosis in an older child. A trend towards significance was found for the association between carrier status and subsequent children. Mothers of confirmed or obligate carrier status tended to have additional children whereas mothers of unknown carrier status were less likely to have additional children after diagnosis of the oldest male. Reports on the effect of carrier status for heritable disease on reproductive choices of mothers have been inconsistent with associations reported in some [Zatz, 1983; Meldrum et al., 2007], but not all [Eggers et al., 1999; Knol et al., 2011], studies. Although the findings in this study appear to suggest that knowing carrier status may increase the likelihood of having subsequent children, the approach to determining carrier status makes it difficult to attribute causative associations with these data. Specifically, MRs were reviewed throughout the entire surveillance period; thus, carrier status may not have been determined by the time of the

subsequent pregnancy. Furthermore, pregnancies subsequent to the oldest affected male could increase the certainty of the mother's carrier status (e.g., a second affected male prompted genetic testing). If the mother did not have any additional children, then carrier status may be more likely to be classified as unknown due to the absence of additional confirmatory information.

Although additional studies have reported that receipt of genetic counseling is associated with reproductive choice [Abramovsky et al., 1980; Zatz, 1983; Rona et al., 1994], no such association was found in the current study. As with carrier status, it is important to note that the findings related to genetic counseling in the current study are limited because of the inability to determine when counseling occurred or the type of information provided in counseling sessions from the MD STAR*net* data. Additionally, information on perception of disease risk, concerns about the effects of an affected child on one's social and personal life, problems caring for the child, and approval or disapproval of relatives were not available for analyses in the MD STAR*net* data.

In summary, findings showed that the reproductive patterns for most women studied did not change after a first diagnosis of DBMD in an affected male child, and may not differ much from those of women in the general US population. The current study is limited by its retrospective design, which did not allow assessment of changes in genetic testing and counseling or prenatal testing that have taken place over the years. Also, the use of secondary data (MRs and BCs) increased the potential for missing or incomplete data, which limited available sample size for analysis [Kirby and Malnory, 1999]. Additionally, stillbirths and terminations were not examined. Nonetheless, the current study is one of the few studies using a population-based sample, which make the findings more generalizable. Further, the study linked MD STAR*net* and BC data, which provided a more complete ascertainment of maternal reproductive history. Future studies should examine more fully the impact of genetic counseling on mothers' future reproductive choices.

Acknowledgments

The authors acknowledge the efforts of all the abstractors, study coordinators, and data managers for their work in data collection and cleaning. This work was funded by grants (U01DD000189 and U01DD000831) sponsored by the Centers for Disease Control and Prevention.

Grant sponsor: Centers for Disease Control and Prevention; Grant numbers: U01DD000189, U01DD000831.

References

Abramovsky I, Godmilow L, Hirschhorn K, Smith H Jr. Analysis of a follow-up study of genetic counseling. Clin Genet. 1980; 17:1–12. [PubMed: 7389180]

American Society of Reproductive Medicine. Age-related fertility decline: A committee opinion. Fertil Steril. 2008; 90:486–487. [PubMed: 18847603]

Brotto LA, Chou AY, Singh T, Woo JS. Reproductive health practices among Indian, Indo-Canadian, Canadian East Asian, and Euro-Canadian women: The role of acculturation. J Obstet Gynaecol Can. 2008; 30:229–238. [PubMed: 18364100]

Bushby K, Finkel R, Birnkrant DJ, Case LE, Clemens PR, Cripe L, Kaul A, Kinnett K, McDonald C, Pandya S, Poysky J, Shapiro F, Tomezsko J, Constantin C. DMD Care Considerations Working Group. Diagnosis and management of Duchenne muscular dystrophy, part 1: Diagnosis, and

pharmacological and psychosocial management. Lancet Neurol. 2010a; 9:77–93. [PubMed: 19945913]

Bushby K, Finkel R, Birnkrant DJ, Case LE, Clemens PR, Cripe L, Kaul A, Kinnett K, McDonald C, Pandya S, Poysky J, Shapiro F, Tomezsko J, Constantin C. DMD Care Considerations Working Group. Diagnosis and management of Duchenne muscular dystrophy, part 2: Implementation of multidisciplinary care. Lancet Neurol. 2010b; 9:177–189. [PubMed: 19945914]

Centers for Disease Control and Prevention (CDC). Prevalence of Duchenne/Becker muscular dystrophy among males ages 5–24 years—Four states, 2007. MMWR. 2009; 58:1119–1122. [PubMed: 19834452]

Ciafaloni E, Fox DJ, Pandya S, Westfield CP, Puzhankara S, Romitti PA, Mathews KD, Miller TM, Matthews DJ, Miller LA, Cunniff C, Druschel CM, Moxley RT. Delayed diagnosis in Duchenne muscular dystrophy: Data from the muscular dystrophy surveillance, tracking, and research network (MD STAR*net*). J Pediatr. 2009; 155:380–385. [PubMed: 19394035]

Eggers S, Pavanello RC, Passos-Bueno MR, Zatz M. Genetic counseling for childless women at risk for Duchenne muscular dystrophy. Am J Med Genet. 1999; 86:447–453. [PubMed: 10508987]

Frets PG, Verhage F, Niermeijer MF. Characteristics of the post-counseling reproductive decision-making process: An explorative study. Am J Med Genet. 1991; 40:298–303. [PubMed: 1951432]

Gardner-Medwin D, Bundey S, Green S. Early diagnosis of Duchenne muscular dystrophy. Lancet. 1978; 1:1102.

Godley J. Kinship networks and contraceptive choice in Nang Rong, Thailand. Int Fam Plan Perspect. 2001; 27:4–10.

Hamilton BE, Ventura SJ. Fertility and abortion rates in the United States, 1960–2002. Int J Androl. 2006; 29:34–45. [PubMed: 16466522]

Hamilton BE, Martin JA, Ventura SJ. Births: Preliminary data for 2005. Natl Vital Stat Rep. 2006; 55:1–18.

Hildes E, Jacobs HK, Cameron A, Seshia SS, Booth F, Evans JA, Wrogemann K, Greenberg CR. Impact of genetic counseling after neonatal screening for Duchenne muscular dystrophy. J Med Genet. 1993; 30:670–674. [PubMed: 8411054]

Hutton EM, Thompson MW. Carrier detection and genetic counseling in Duchenne muscular dystrophy: A follow-up study. Can Med Assoc J. 1976; 115:749–752. [PubMed: 974964]

Kirby RS, Malnory M. Limitations of databases. Pediatrics. 1999; 103:531–532. [PubMed: 10026064]

Knol HM, Voskuilen MA, Holterman F, Kluin-Nelemans JC, Meijer K. Reproductive choices and obstetrical experience in Dutch carriers of haemophilia A and B. Haemophilia. 2011; 17:233–236. [PubMed: 21332882]

Maternowska C, Estrada F, Campero L, Herrera C, Brindis CD, Vostrejs MM. Gender, culture and reproductive decision-making among recent Mexican migrants in California. Cult Health Sex. 2010; 12:29–43. [PubMed: 19657804]

Mathews KD, Cunniff C, Kantamneni JR, Ciafaloni E, Miller T, Matthews D, Cwik V, Druschel C, Miller L, Meaney FJ, Sladky J, Romitti PA. Muscular dystrophy surveillance tracking and research network (MD STAR*net*): Case definition in surveillance for childhood-onset Duchenne/Becker muscular dystrophy. J Child Neurol. 2010; 25:1098–1102. [PubMed: 20817884]

Meldrum C, Scott C, Swoboda KJ. Spinal muscular atrophy genetic counseling access and genetic knowledge: Parents' perspectives. J Child Neurol. 2007; 22:1019–1026. [PubMed: 17761658]

Miller LA, Romitti PA, Cunniff C, Druschel C, Mathews KD, Meaney FJ, Matthews D, Kantamneni J, Feng ZF, Zemblidge N, Miller TM, Andrews J, Fox D, Ciafaloni E, Pandya S, Montgomery A, Kenneson A. The muscular dystrophy surveillance tracking and research network (MD STAR*net*): Surveillance methodology. Birth Defects Res A Clin Mol Teratol. 2006; 76:793–797. [PubMed: 17036307]

Oladeji D. Socio-cultural and norms factors influencing family planning choices among couples in Ibadan Metropolis, Nigeria. Eur J Sci Res. 2008; 23:212–218.

Parsons EP, Clarke AJ, Hood K, Lycett E, Bradley DM. Newborn screening for Duchenne muscular dystrophy: A psychosocial study. Arch Dis Child Fetal Neonatal Ed. 2002; 86:F91–F95. [PubMed: 11882550]

Raymond FL, Whittaker J, Jenkins L, Lench N, Chitty LS. Molecular prenatal diagnosis: The impact of modern technologies. Prenat Diagn. 2010; 30:674–681. [PubMed: 20572117]

Romitti PA, Watanabe-Galloway S, Budelier WT, Lynch CF, Puzhankara S, Wong-Gibbons D, Hoppin JA, Alavanja MC. Identification of Iowa live births in the agricultural health study. Arch Environ Occup Health. 2010; 65:154–162. [PubMed: 20705576]

Rona RJ, Beech R, Mandalia S, Donnai D, Kingston H, Harris R, Wilson O, Axtell C, Swan AV, Kavanagh F. The influence of genetic counseling in the era of DNA testing on knowledge, reproductive intentions and psychological wellbeing. Clin Genet. 1994; 46:198–204. [PubMed: 7820931]

Smith RA, Sibert JR, Wallace SJ, Harper PS. Early diagnosis and secondary prevention of Duchenne muscular dystrophy. Arch Dis Child. 1989; 64:787–790. [PubMed: 2476078]

Wertz DC, Sorenson JR, Heeren TC. Genetic counseling and reproductive uncertainty. Am J Med Genet. 1984; 18:79–88. [PubMed: 6741997]

Zatz M. Effects of genetic counseling on Duchenne muscular dystrophy families in Brazil. Am J Med Genet. 1983; 15:483–490. [PubMed: 6881214]

TABLE I

Distribution of Selected Maternal Characteristics by Study Inclusion Status

	Included mothers (N = 239)		Excluded mothers (N = 72)		
Maternal Characteristic	**Mean (SD)**	**Range**	**Mean (SD)**	**Range**	***P*-value**
Age[a] (years)	26.2 (5.2)	15.0–44.0	25.9 (6.1)	14.0–39.0	0.75
	n	%	n	%	
Race/ethnicity					<0.01
White non-Hispanic	151	74.8	24	48.0	
All other[b]	51	25.2	26	52.0	
Education[a]					0.76
High school or less	103	54.2	18	60.0	
Some college	52	27.4	8	26.7	
Bachelor's degree or higher	35	18.4	4	13.3	
Parity[a]					0.19
0	79	33.1	10	20.4	
1	93	38.9	31	42.9	
2 or more	67	28.0	18	36.7	
Knowledge of DBMD family history[a,c]					0.61
Known history	62	25.9	11	22.4	
No known history	177	74.1	38	77.6	
Carrier status[d]					<0.01
Suspected carrier	9	3.8	8	11.1	
Carrier	115	48.1	24	33.3	
Non-carrier	52	21.8	12	16.7	
Unknown carrier	63	26.4	28	38.9	
Genetic counseling[d]					<0.01
Recommended	17	7.1	8	11.1	
Received	166	69.5	33	45.8	
Unknown	56	23.4	31	43.1	

DBMD, Duchenne or Becker muscular dystrophy; SD, standard deviation.

Overall N may not equal to total n because of missing values.

[a] At time of delivery of oldest affected male child.

[b] Includes Hispanic, black non-Hispanic, Asian/Pacific islander, native American, and mixed race.

[c] Excludes all mothers with undetermined family history.

[d] At any time point prior to completion of surveillance data collection in 2008.

TABLE II

Association Between Selected Maternal Characteristics and One or More Live Births After Diagnosis of Oldest Affected Male Child

	Live birth after diagnosis of oldest affected male child				
	Yes (N = 43)		No[a] (N = 196)		
Maternal characteristic	**Mean (SD)**	**Range**	**Mean (SD)**	**Range**	***P*-value**
Age[b]	26.2 (4.2)	19.0–40.0	31.5 (5.5)	16.0–46.0	<0.01
	n	%	n	%	
Race/ethnicity					<0.01
White non-Hispanic	23	57.5	128	79.0	
All other[c]	17	42.5	34	21.0	
Education[d]					0.78
High school or less	20	58.8	83	53.1	
Some college	9	26.5	43	27.6	
Bachelor's degree or higher	5	14.7	30	19.2	
Parity[b]					0.44
1	12	27.9	38	19.4	
2	16	37.2	77	39.3	
3 or more	15	34.8	81	41.3	
Knowledge of DBMD family history[d]					0.48
Known history	13	30.2	49	25.0	
No known history	30	69.8	147	75.0	
Carrier status[e]					0.06
Confirmed or obligate[f]	26	60.5	92	46.9	
Non-carrier	11	25.6	41	20.9	
Unknown carrier[g]	6	14.0	63	32.1	
Genetic counseling[e]					0.50
Recommended	2	4.7	15	7.6	
Received	33	79.7	133	67.9	
Unknown	8	18.6	48	24.5	

DBMD, Duchenne or Becker muscular dystrophy; SD, standard deviation.

Overall N may not equal to total n because of missing values.

[a] Includes all mothers who had no live birth after delivery of oldest affected male (n = 143) and mothers who had a live birth before diagnosis of oldest affected male (n = 53).

[b] At time of diagnosis of oldest affected male child.

[c] Includes Hispanic, black non-Hispanic, Asian/Pacific islander, native American, and mixed race.

[d] At time of delivery of oldest affected male child.

[e] At any time point prior to completion of surveillance data collection in 2008.

[f] Includes all mothers with known family history of DBMD who were classified as suspected carriers (n = 3).

[g] Includes all mothers with no known family history of DBMD who were classified as suspected carriers (n = 6).

Am J Med Genet A. Author manuscript; available in PMC 2018 April 06.